





Test No.	Test Name	Price	Test No.	Test Name	Price
	<b>Glycogen Storage Disease</b>				
220	GYS2 Sequencing (Type 0)	\$ 940			
221	G6PC Sequencing (Type Ia)	\$ 440	172	MY7 Sequencing	\$ 1640
222	SLC37A4 Sequencing (Type Ib)	\$ 540	175	MYL2 Sequencing	\$ 480
223	GAA Sequencing (Type II)	\$ 990	176	MYL3 Sequencing	\$ 450
224	AGL Sequencing (Type III)	\$1,790	178	TNNI3 Sequencing	\$ 570
225	GBE1 Sequencing (Type IV)	\$ 990	179	TNNT2 Sequencing	\$ 820
226	PYGM Sequencing (Type V)	\$340 - 990	177	TPM1 Sequencing	\$ 620
227	PYGM Sequencing (Type V) – Tier 1 only	\$ 340	144	VCL Sequencing	\$1,170
228	PYGM Sequencing (Type V) – Tier 2 only	\$ 790			
229	PYGL Sequencing (Type VI)	\$1,140			
230	PFKM Sequencing (Type VII)	\$1,090		<b>Inclusion Body Myopathy</b>	
231	PHKA1 Sequencing (Type IX)	\$1,690	367	GNE Sequencing	\$ 740
232	PHKA2 Sequencing (Type IX)	\$490–1,690	361	MYH2 Sequencing	\$1,690
233	PHKB Sequencing (Type IX)	\$1,690		<b>Isovaleric Acidemia</b>	
235	PHKG2 Sequencing (Type IX)	\$ 490	250	IVD Sequencing	\$ 590
				<b>Joubert Syndrome</b>	
			260	Joubert Syndrome Sequential Test includes: AHI1, CEP290-Tier 1, MKS3, NPHP1 Deletion Test	\$940-3,330
	<b>Glycyl tRNA Synthetase-Related Disorders</b>		270	AHI1 Sequencing	\$1,390
383	GARS Sequencing	\$ 940	277	ARL13B Sequencing	\$ 640
	<b>GM1 Gangliosidosis/Morquio Syndrome Type B</b>		278	CC2D2A Sequencing	\$1,590
477	GLB1 Sequencing	\$ 840	271	CEP290 Sequencing	\$640-2,190
			272	CEP290 Sequencing – Tier 1 only	\$ 640
			273	CEP290 Sequencing – Tier 2 only	\$1,690
	<b>Heat Shock 22/27 kDa Protein-Related Disorders</b>		274	TMEM67/MKS3 Sequencing	\$1,290
387	HSPB1 Sequencing	\$ 390	275	NPHP1 Deletion Testing	\$ 290
389	HSPB8 Sequencing	\$ 440	651	NPHP1 Sequencing	\$1,090
			276	RPGRIP1L Sequencing	\$1,190
			291	TMEM216 Sequencing	\$ 460
	<b>Holoprosencephaly</b>				
580	Holoprosencephaly Sequential (SHH, ZIC2, SIX3, TGIF1, GLI2)	\$390-3,290			
586	GLI2 Sequencing	\$ 990		<b>Joubert and Meckel-Gruber Syndromes</b>	
585	PTCH1 Sequencing	\$1,090	278	CC2D2A Sequencing	\$1,590
581	SHH Sequencing	\$ 470		<b>Krabbe Disease</b>	
583	SIX3 Sequencing	\$ 440	631	GALC Sequencing	\$ 830
584	TGIF1 Sequencing	\$ 390	632	GALC Deletion Test	\$ 190
582	ZIC2 Sequencing	\$ 540		<b>Leber Congenital Amaurosis</b>	
	<b>3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency</b>		681	CRB1 Sequencing	\$1,040
240	HMGCL Sequencing	\$ 590	665	CRX Sequencing	\$ 440
				<b>Lymphedema</b>	
			280	FOXC2 Sequencing	\$ 390
			281	FLT4 Sequencing – Tier 1 only	\$ 540
	<b>Hypertrophic Cardiomyopathy</b>		282	FLT4 Sequencing – Tier 2 only	\$ 950
190	Sequential Testing of Eight Sarcomeric Genes	\$5,580	283	SOX18 Sequencing	\$ 490
191	MYBPC3-MYH7 Sequencing	\$2,630		<b>Lysosomal Storage Disorders</b>	
193	MYL2-MYL3-ACTC1 Sequencing	\$1,240	477	GLB1 Sequencing	\$ 840
192	TNNT2-TNNI3-TPM1 Sequencing	\$1,710	476	HEXB Sequencing – Tier 1 only	\$ 840
174	ACTC1 Sequencing	\$ 530		<b>Malignant Hyperthermia /Central Core Disease</b>	
198	ACTN2 Sequencing	\$1,090	570	Sequential RYR1 Sequencing	\$740–3,990
196	CSRP3 Sequencing	\$ 470	571	RYR1 Sequencing – Tier 1 only	\$ 740
			572	RYR1 Sequencing – Tier 2 only	\$3,790

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			366	MYOT/TTID Sequencing	\$ 190-590
			355	NEB Sequencing	\$3245-6490
	<b>Meckel Gruber Syndrome</b>		356	NEB Exon 55 Deletion Test	\$ 200
301	MKS1 Sequencing	\$290 - 840	330	SEPN1 Sequencing	\$ 690
302	MKS1 Sequencing – Tier 1 only	\$ 290	348	POMT1 Sequencing	\$ 890
303	MKS1 Sequencing – Tier 2 only	\$ 790	349	POMT2 Sequencing	\$ 990
276	RPGRIP1L Sequencing	\$1,190	351	POMGNT1 Sequencing	\$ 640
274	TMEM67/MKS3 Sequencing	\$1,290	352	SGCA Sequencing	\$ 490
	<b>Mental Retardation</b>		353	SGCB Sequencing	\$ 440
561	CUL4B Sequencing	\$1,090	338	SGCG Sequencing	\$ 570
562	SLC9A6 Sequencing	\$ 790	246	SYNE1 Sequencing	\$6,490
134	VPS13B Sequencing	\$2,790	247	SYNE1 Seq. - French Canadian Mutation Panel	\$ 550
	<b>Metachromatic Leukodystrophy</b>		468	TCAP Sequencing	\$ 390
511	PSAP Sequencing	\$ 790	385	TTN Sequencing	\$ 1290
	<b>Methylmalonic Acidemia</b>			<b>Myofibrillar Myopathy</b>	
310	Sequential Gene Sequencing	\$740-1640	362	CRYAB Sequencing	\$ 370
311	MUT Sequencing	\$ 740	363	DES Sequencing	\$ 670
312	MMAA Sequencing	\$ 490	364	FLNC Sequencing	\$2,490
313	MMAB Sequencing	\$ 540	365	LDB3 (ZASP) Sequencing	\$ 940
	<b>3-Methylcrotonyl-CoA Carboxylase Deficiency</b>			<b>Multiple Sulfatase Deficiency</b>	
320	Sequential Gene Sequencing	\$840-1,770	512	SUMF1 Sequencing	\$ 590
321	MCCC2 Sequencing	\$ 840		<b>Myotilinopathy</b>	
322	MCCC1 Sequencing	\$ 990	366	MYOT/TTID Sequencing	\$ 190-590
	<b>3-Methylglutaconic Aciduria Type I</b>			<b>Nemaline Myopathy</b>	
323	AUH Sequencing	\$ 690	358	ACTA1 Sequencing	\$ 530
	<b>Mucopolipidosis</b>		355	NEB Sequencing	\$3245-6490
316	GNPTAB Sequencing	\$ 1,190	356	NEB Exon 55 Deletion Test	\$ 200
317	GNPTG Sequencing	\$ 470	331	TPM2 Sequencing	\$ 590
	<b>Multiminicore, Rigid Spine Muscular Dystrophy</b>		357	TPM3 Sequencing	\$ 570
330	SEPN1 Sequencing	\$ 690	332	TNNT1 Sequencing	\$ 690
357	TPM3 Sequencing	\$ 570		<b>Nephronophthisis</b>	
	<b>Multiple Carboxylase Deficiency</b>				
520	BTD Sequencing	\$ 540	656	GLIS2/NPHP7 Deletion Test	\$ 540
521	HLCS Sequencing	\$ 740	652	INVS/NPHP2 Deletion Test	\$ 940
	<b>Multiple Pterygium Syndrome</b>		655	IQCB1/NPHP5 Deletion Test	\$ 740
415	CHRNA1, CHRND, CHRNG Sequencing	\$680-1890	275	NPHP1 Deletion Test	\$ 290
405	CHRNG Sequencing	\$ 730	651	NPHP1 Sequencing	\$1,090
	<b>Multiple Sulfatase Deficiency</b>		653	NPHP3 Sequencing	\$1,320
512	SUMF1 Sequencing	\$ 590	654	NPHP4 Sequencing	\$1,350
	<b>Muscular Dystrophy</b>			<b>Neurofibromatosis</b>	
340	Dyostroglycanopathy Sequential (FKRP, FKTN, POMGnT1, LARGE1)	\$330 - 3730	117	NF1 Sequencing	\$2,490
341	CAPN3 Sequencing	\$1,090	118	NF2 Sequencing	\$ 840
467	CAV3 Sequencing	\$ 390	116	SPRED1 Sequencing	\$ 540
384	DAG1 Sequencing	\$ 690		<b>Neurofibromatosis-Noonan Syndrome</b>	
465	DOK7 Sequencing	\$ 540	115	NF1 Sequencing	\$540-2490
342	DYSF Sequencing	\$2,490		<b>Niemann-Pick Disease</b>	
344	FKRP Sequencing	\$ 390	473	NPC1 Sequencing	\$1,240
343	FKTN /FCMD Sequencing	\$ 590	474	NPC2 Sequencing	\$ 490
244	ITGA7 Sequencing	\$1,240		<b>Noonan Syndrome/LEOPARD Syndrome</b>	
345	LAMA2 Sequencing	\$2,990	370	Sequential (PTPN11, SOS1, RAF1, KRAS)	\$390 - 1,990
245	LAMA2: Mexican Exon 55 Mutation	\$ 190	371	Sequential – Tier 1 only	\$ 390
346	LARGE Sequencing	\$ 890	372	Sequential – Tier 2 only	\$1,990
347	LMNA Sequencing	\$ 590	373	Noonan PTPN11 Sequencing	\$ 390 - 890

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371	Noonan <i>PTPN11</i> Sequencing – Tier 1 only	\$ 390			
374	Noonan <i>PTPN11</i> Sequencing – Tier 2 only	\$ 690			
375	<i>RAF1</i> Sequencing	\$ 370			
376	<i>SOS1</i> Sequencing	\$590 - 1,190	478	<b>Tay-Sachs Disease</b>	
377	LEOPARD <i>PTPN11</i> Sequencing	\$370 - 890	475	<i>GM2A</i> Sequencing	\$ 440
378	<i>KRAS</i> Sequencing	\$ 440		<i>HEXA</i> Sequencing	\$ 770
115	<i>NF1</i> Sequencing	\$540-2490			
	<b>Pallister-Hall/Greig Cephalopolysyndactyly</b>		430	<b>Thrombocytopenia</b>	
380	<i>GLI3</i> Sequencing	\$1,090	431	<i>ADAMTS13</i> Sequencing	\$1,490
	<b>Pena-Shokeir Syndrome, Type 1</b>		432	<i>GATA1</i> Sequencing	\$ 390
413	<i>DOK7-RAPSN</i> Sequencing Panel	\$540-\$1060	433	Bernard-Soulier Sequential( <i>GP1BA, GP1BB, GP9</i> )	\$490 - 890
	<b>Phenylalanine Hydroxylase Deficiency (PKU)</b>		434	<i>GP1BA</i> Sequencing	\$ 490
530	<i>PAH</i> Testing	\$ 690	435	<i>GP1BB</i> Sequencing	\$ 390
			436	<i>GP9</i> Sequencing	\$ 340
	<b>Progeria, Hutchinson-Gilford</b>		438	<i>MASTL</i> Sequencing	\$ 740
347	<i>LMNA</i> Sequencing	\$ 590	437	<i>MYH9</i> Sequencing	\$ 540
	<b>Propionic Acidemia</b>		439	<i>MPL</i> Sequencing	\$ 640
390	Sequential Gene Sequencing	\$740-1,890	440	<i>RUNX1</i> Sequencing	\$ 490
391	<i>PCCA</i> Sequencing	\$1,290		<i>WAS</i> Sequencing	\$ 540
392	<i>PCCB</i> Sequencing	\$ 790			
	<b>Rapsyn-Related Disorders</b>				
466	<i>RAPSN</i> Sequencing	\$ 590	541	<b>Tyrosinemia</b>	
				<i>FAH</i> Sequencing	\$ 690
	<b>Restrictive Dermopathy, Mandibuloacral Dysplasia</b>		359	<b>Ullrich Congenital Muscular Dystrophy</b>	
400	<i>ZMPSTE24</i> Sequencing	\$ 690		<i>COL6A1-COL6A2-COL6A3</i> Sequencing	\$1290-3890
	<b>Robinow Syndrome, Autosomal Recessive</b>				
136	<i>ROR2</i> Sequencing	\$ 740			
			552	<b>Urea Cycle Disorders</b>	
	<b>Sandhoff Disease</b>		553	<i>ARG1</i> Sequencing	\$ 490
476	<i>HEXB</i> Sequencing	\$ 840	554	<i>ASS1</i> Sequencing	\$ 740
	<b>Seipin-Related Disorders</b>		555	<i>CPS1</i> Sequencing	\$1,890
461	<i>BSCL2</i> Sequencing	\$ 740	551	<i>NAGS</i> Sequencing	\$ 430
	<b>Sialuria</b>			<i>OTC</i> Sequencing	\$ 540
368	<i>GNE</i> , Exon 5 Sequencing	\$ 190			
	<b>Simpson-Golabi-Behmel Syndrome</b>		450	<b>Ventricular Tachycardia</b>	
563	<i>GPC3</i> Sequencing	\$ 590		<i>RYR2</i> Sequencing	\$1,390
			641	<b>Vision Disorders</b>	
	<b>Skeletal Disorders</b>			<i>COL18A1</i> Sequencing	\$1,990
420	Sequential Test ( <i>DLL3, MESP2, LFNG</i> )	\$540-1,290	610	<b>White Matter Disorders</b>	
421	<i>DLL3</i> Sequencing	\$ 590		<i>EIF2B1 – EIF2B5</i> Sequential Sequencing	\$690-2,390
424	<i>GDF6</i> Sequencing	\$ 490	460	<b>Wilson Disease</b>	
422	<i>LFNG</i> Sequencing	\$ 490		<i>ATP7B</i> Sequencing	\$ 990
423	<i>MESP2</i> Sequencing	\$ 390			
	<b>Smith-Lemli-Opitz Syndrome</b>				
410	<i>DHCR7</i> Sequencing	\$ 440			
	<b>Sotos Syndrome</b>		500	<b>- Other Tests -</b>	
132	<i>NSD1</i> Sequencing	\$1,490		<b>DNA Banking</b>	\$ 98
	<b>Sphingolipidoses</b>		470		
620	<i>ARSA</i> Sequencing	\$ 490	490	<b>Sex Chromosome Panel</b>	\$ 90
479	<i>GBA</i> Sequencing	\$ 630		<b>DNA Identity Panel</b>	\$ 90
	<b>Spinal Muscular Atrophy with Respiratory Distress Type 1</b>			Gender Analysis, Zygosity Testing, Specimen Matching	
339	<i>IGHMBP2</i> Sequencing	\$ 910		<b>Genome Polymorphism Scan</b>	\$ 590